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Bone scinitigraphy in rhabdomyolysis associated with carnitine palmitoyl transferase deficiency

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A 15-year-old athletic male was hospitalized with back pain and myalgia after standing out in the cold weather watching a soccer match. He was passing dark red urine on admission. He took no prescribed or illicit medications. Family history and physical examination was unremarkable. Laboratory analysis revealed a creatine kinase of 113 542 U/L and normal renal function.

A diagnosis of rhabdomyolysis was made and the patient was treated with intravenous fluids. The cause was thought to be excessive exercise given his athletic background; however, bone scintigraphy revealed diffuse symmetrical isotope uptake in the gluteal muscles, quadriceps, calves and latissimus dorsi (Figures 1 and 2) suggesting non-traumatic aetiology. On closer questioning, the patient had suffered repeated episodes of muscle cramps usually after a period of reduced calorific intake since the age of 11. The history and imaging findings suggested an underlying metabolic defect. Skin biopsy confirmed a diagnosis of carnitine palmitoyl-CoA transferase type II (CPT-II) deficiency. CPT-II enzyme is responsible for the hydrolysis of long-chain fatty acids bound to carnitine following transport across the inter-mitochondrial membrane. Muscle CPT-II deficiency leads to failure of fatty acid oxidation resulting in rhabdomyolysis following conditions including fasting, cold exposure and sleep deprivation.

Rhabdomyolysis can be directly visualized due to isotope deposition in damaged myocytes. The mechanism may be related to isotope adsorption to intracellular calcium released from sarcolemmal disruption [1]. Traumatic and non-traumatic rhabdomyolysis can be difficult to distinguish when confounding factors exist, particularly in alcoholics and drug users. Bone scintigraphy usually shows asymmetrical, localized tracer uptake in traumatic lesions and a more diffuse, symmetrical pattern in non-traumatic rhabdomyolysis [2]. Identifying non-traumatic rhabdomyolysis is important as there is usually an underlying precipitant that needs to be removed before recovery can take place. Bone scintigraphy can distinguish between non-traumatic and traumatic rhabdomyolysis by the pattern of isotope uptake in damage myocytes.

Conflict of interest statement. None declared.

References

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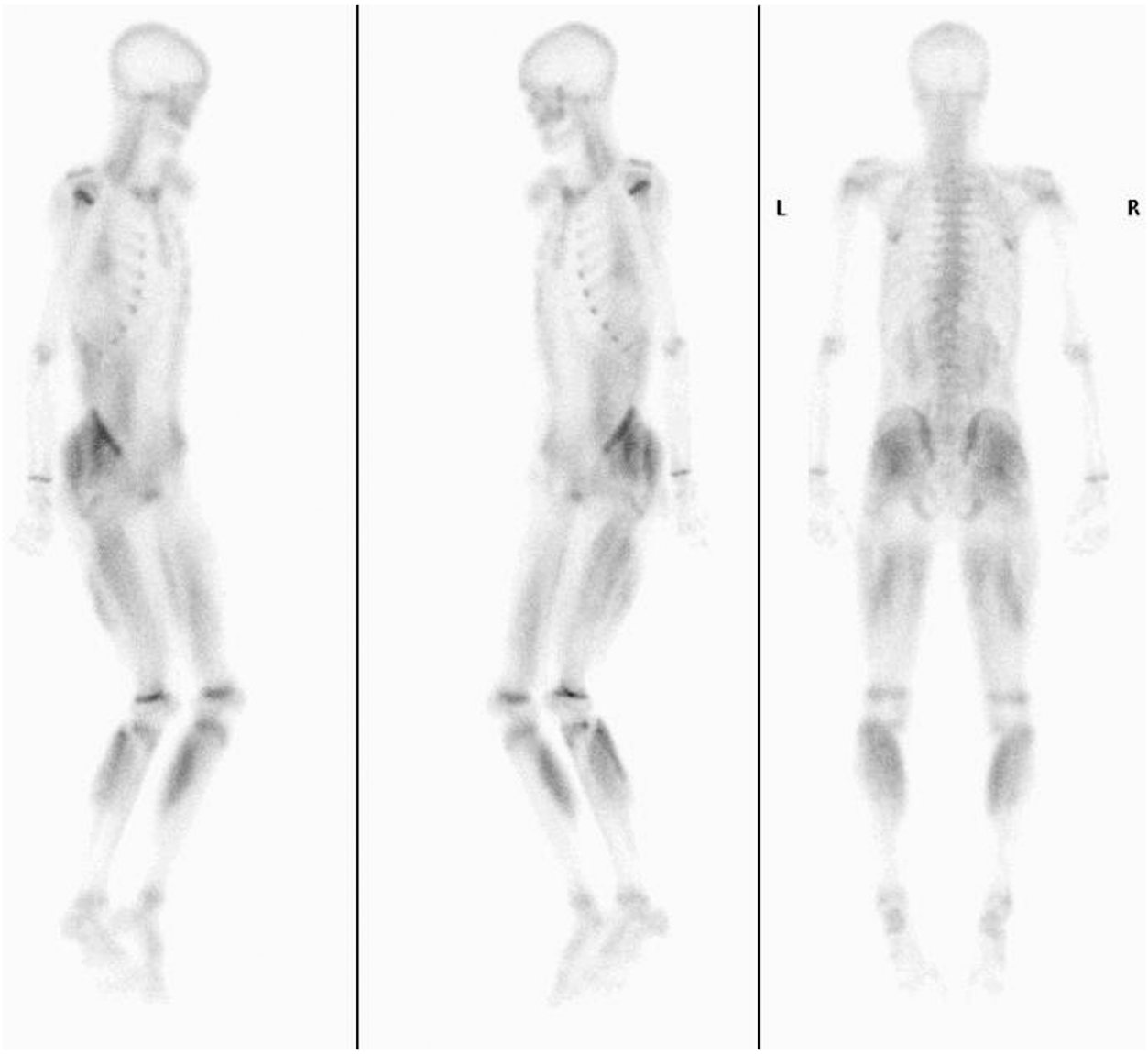

Fig. 1. Isotope bone scan using 561 MBq of technetium 99-labelled methylene diphosphonate showed marked muscular uptake in the gluteal muscles, quadriceps, calf muscles and latissimus dorsi.

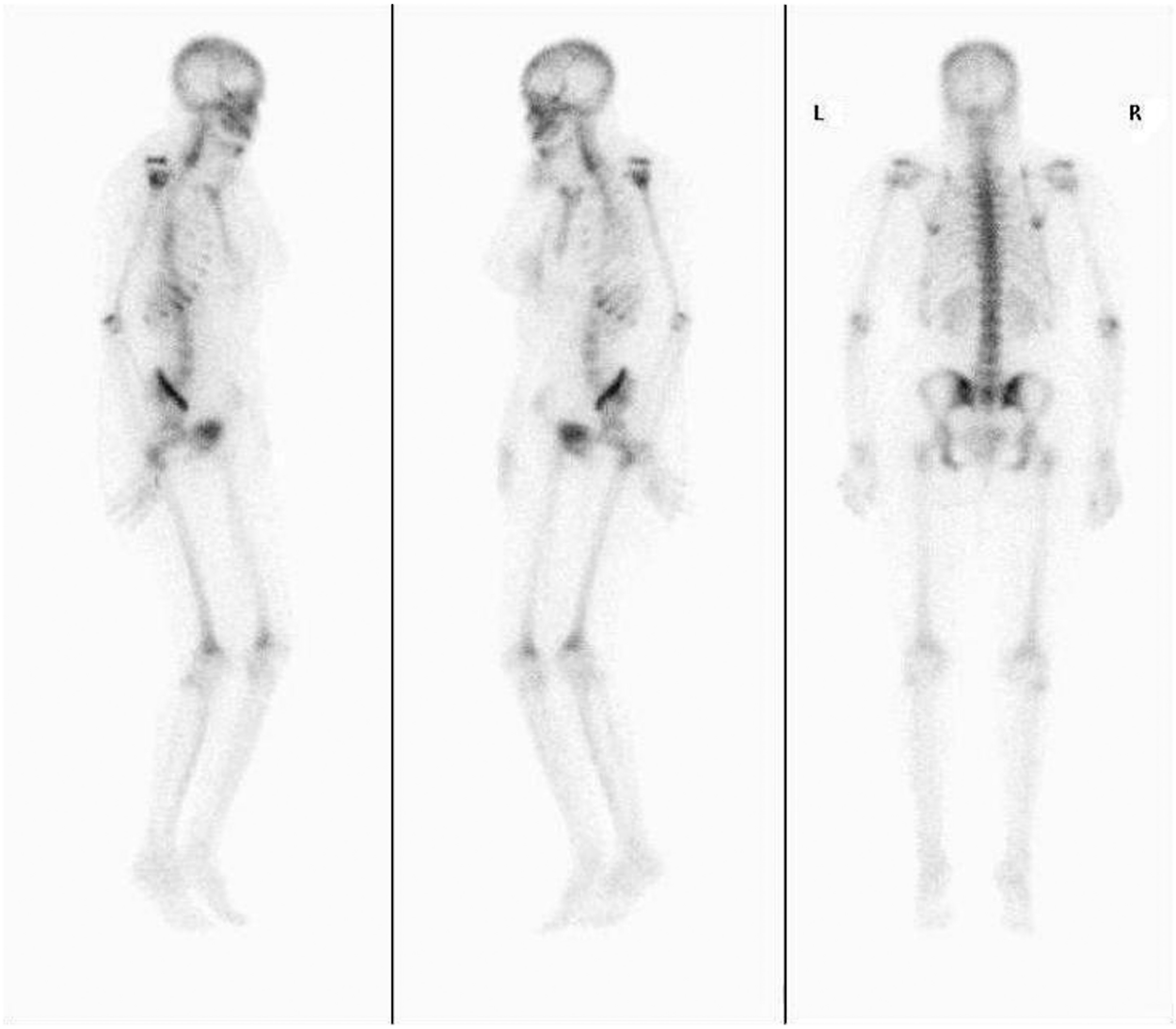

Fig. 2. Normal isotope bone scan for comparison, no muscular isotope deposition is visible.